

Table S1. Tissue-specific m.3243A>G mutation load and blood lactate and pyruvate levels of a boy born following PGD selection to a mother with MELAS.

Proband age at testing	m.3243A>G mutation level in tissues (detection method)				Lactate levels		Pyruvate levels	Lactate:Pyruvate ratio
	Trophectoderm	Blood	Urine	Buccal	Blood	Plasma	Blood	Blood
Preimplantation blastocyst	12% (qPCR)*							
Birth				15% (method not reported)*				
1.5-months		47% (ARMS-qPCR)	52% (ARMS-qPCR)		2.3 mM (normal <2 mM)		0.10 mM (normal <0.14 mM)	23 (normal 10-20)
2.5-month					1.6 mM (normal <2 mM)		0.08 mM (normal <0.14 mM)	20 (normal 10-20)
3-months					2.3 mM (normal <1.6 mM)	1.8 mM (normal <2 mM)		
10-months						1.8 mM (normal <2 mM)		
14-months						1.7 mM (normal <2 mM)		
16-months**					3.5 mM (normal <1.6 mM)			
18-months		46% (ARMS-qPCR)	42% (ARMS-qPCR)		1.3 mM (normal <2 mM)		0.09 mM (normal <0.14 mM)	14 (normal 10-20)
33-months					1.4 mM (normal <2 mM)		0.08 mM (normal <0.14 mM)	18 (normal 10-20)

* qPCR, quantitative PCR, as reported by Treff et al., *Fertility and Sterility*, 2012

**, intercurrent febrile illness

ARMS-qPCR - amplification refractory mutation system quantitative PCR